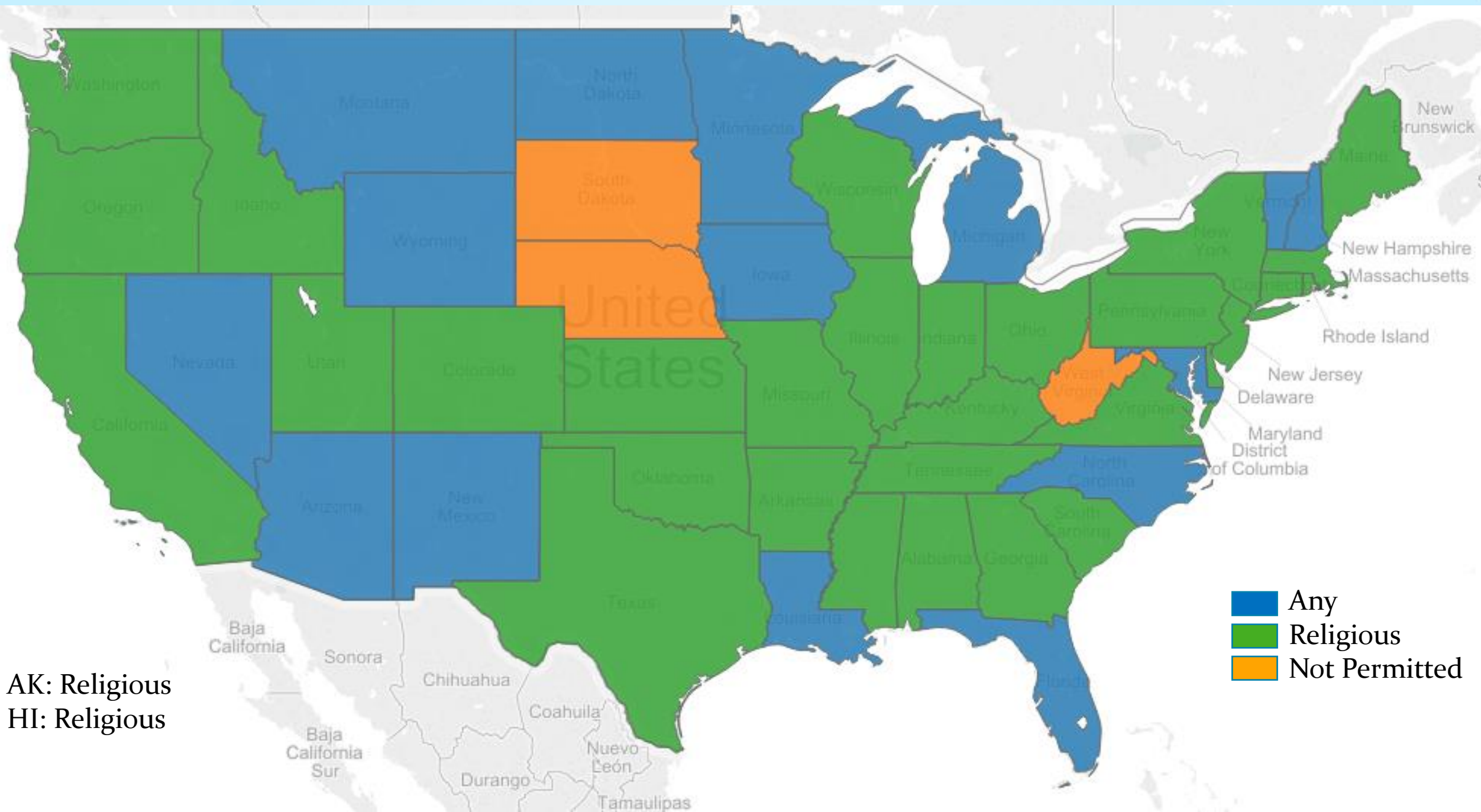


# Understanding Newborn Screening Refusals

JEREMY PENN, PH.D.





- Any
- Religious
- Not Permitted

	Number of States	Percent of States
Refusal for any reason	15	29%
Refusal for religious reasons	33	65%
No provision for refusals	3	6%

Penn JD, Sondreal E. Review of best practices in documenting newborn screening refusals for states. Paper presented at the 2014 APHL Newborn Screening and Genetic Testing Symposium: Newborn Screening: Re-Assessing Business as Usual. Anaheim, CA.

# North Dakota Newborn Screening Refusal Law

## **33-06-16-04. Refusal of testing.**

1. If the parents or guardians refuse to have their infant receive newborn screening testing after being provided written information, that refusal shall be documented by a written statement signed by the parents or guardians.
2. The original refusal statement shall become a part of the infant's medical record and a copy of the statement must be submitted to the newborn screening program within six days after testing was refused.

# The “Best Interest” Standard

Courts have often required states to “establish that parental choices endanger the child and thus fall below the acceptable threshold. In general, courts have gone against parents when the life of a child is endangered, but have typically given great discretion to parents in situations that are not imminently life-threatening.”

(Diekema DS. Parental refusals of medical treatment: the harm principle as threshold for state intervention. *Theor Med Bioeth.* 2004;25(4), pp. 248-249)

# For example

- For a child with cancer, parents would have the right to make decisions about which treatment(s) are in the “best interest” of the child – parental rights are protected in most circumstances
- However, in some instances states have compelled children to undergo chemotherapy even when it was against the desires of the parents and the child (when the life of the child was endangered)
- How does this principle apply to newborn screening?
- If a child has, say, SCID, clearly screening is in the child’s best interest. But what of the many who do not have SCID? Does the best interest principle give the state the right to compel screening for all, even against the wishes of parents?

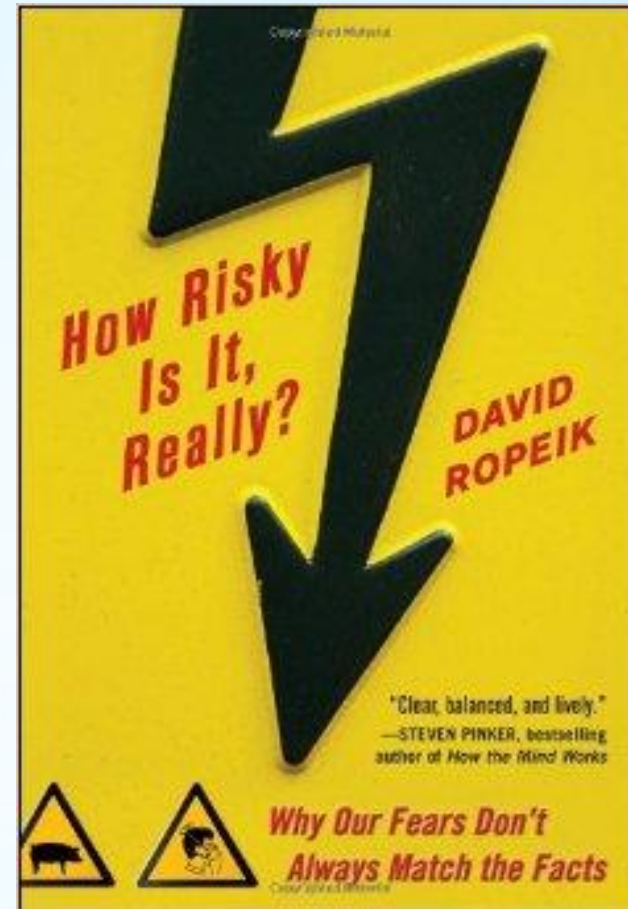


# Risks for Refusing Screening

# *How Risky is it Really?*

Many people are  
bad at estimating  
risk

David Ropeik, 2010





Risk perception gap: A dramatic but small risk can be more influential than a more subtle but larger risk



Many screened  
conditions  
“clinically silent”



# For example

- Most babies with MCAD deficiency (medium chain acyl-CoA dehydrogenase deficiency) look normal at birth
- For some, the first sign of MCAD is a metabolic crisis
  - 20-25% die in this first episode (Dyack, 2005)

Dyack S. Expanded newborn screening: Lessons learned from MCAD deficiency. *Paediatr Child Health*. 2004;9(4): 241-243.

# For example

- Severe Combined Immunodeficiency (to be added to ND's panel later this year)
- Untreated has 100% death rate
- Without family history or screening, first sign of SCID is recurring infections which can be life-threatening
- With early diagnosis through screening and treatment the long-term survival rate can be as high as 94%

(Pai S, Logan BR, Griffith LM, Buckley RH, Parrott RE, Dvorak CC, et al. Transplantation outcomes for severe combined immunodeficiency, 2000-2009. *New England Journal of Medicine*. 2014;371,434-446.)

# Historical Perspective

- Before the expansion of the NBS panel, the risk of refusal was much lower
- Rate of PKU approximately 35 per 100,000 births, or 0.035% chance
- Many birth providers could go a full career and never have a PKU patient



Me, You, and PKU.  
<https://meupku.wordpress.com/2013/06/04/the-power-of-routine/>



# Expanded Newborn Screening

- Increased number of conditions screened
- More benefits to babies through early intervention
- Changes the risk estimate because babies much more likely to have a true positive screen





# How are new conditions added to the screening panel?

- Each state makes its own decision about what conditions to screen and when
- Guidance at the national level from the Advisory Committee on Heritable Disorders in Newborns and Children
- Extensive evidence review with a very high standard for addition of conditions to the recommended uniform screening panel – requires large pilot studies and expert review of the evidence
- Emphasis on evidence that screening will have health benefits for the child
- Conditions for which there is no known treatment are NOT added to the screening panel
- Use the Wilson and Jungner (1968) criteria to inform the decision process

Wilson JMG, Jungner G. Principles and practice of screening for disease.

[http://whqlibdoc.who.int/php/WHO\\_PHP\\_34.pdf](http://whqlibdoc.who.int/php/WHO_PHP_34.pdf). Geneva: World Health Organization; 1968. Accessed March 22, 2015.

Estimated number of babies identified through  
newborn screening each year in the United States:

12,578

317.82 per 100,000 births

Centers for Disease Control and Prevention. CDC grand rounds: Newborn screening and improved outcomes. *Morbidity and Mortality Weekly Report*. 2012 June 1; 61(21): 390-393, plus standard estimates for incidence of CCHD, Pompe, and SCID.

Risk for Refusing Newborn Screening Compared to Other Common Health Risks for Infants				
Health Risk	Number impacted in the United States	Rate per 100,000 <sup>a</sup>	Risk ratio for newborn screening <sup>b</sup>	Risk ratio for bloodspot screening only <sup>c</sup>
All newborn screening <sup>d</sup>	12,578	317.82	1.00	0.54
Bloodspot newborn screening only <sup>e</sup>	6,785	171.44	1.85	1.00
Pertussis death (under age 1) <sup>f</sup>	12	0.30	1048.16	565.41
Death by motor vehicle accident (under age 1) <sup>g</sup>	68	1.72	184.97	99.78
Death from influenza and pneumonia (under age 1) <sup>g</sup>	178	4.50	70.66	38.12
Death from certain intestinal infections <sup>f</sup> (under age 1) <sup>g</sup>	227	5.74	55.41	29.89
Death, all accidents (unintentional injuries including motor vehicle accidents, under age 1) <sup>g</sup>	1,156	29.21	10.88	5.87
Injured in car crash (under age 1) <sup>h</sup>	3,304	83.49	3.81	2.05
All causes of death (under age 1) <sup>g</sup>	23,440	592.28	0.54	0.29

a. Based on 3,957,577 live births in the United States in 2013.

b. Includes estimates for critical congenital heart disease, severe combined immunodeficiency, and Pompe.

c. Includes estimates for severe combined immunodeficiency and Pompe.

d. Yearly average, most victims are children under age 2.

e. Includes A04, A07-A09, such as e. coli, giardiasis, rotaviral enteritis, and infections gastroenteritis.

f. National Center for Immunization and Respiratory Diseases: Division of Bacterial Diseases. 2013 final pertussis surveillance report. <http://www.cdc.gov/pertussis/downloads/pertuss-surv-report-2013.pdf>. Published October, 2014. Accessed October 20, 2015.

g. Centers for Disease Control and Prevention. National Vital Statistics Report. Deaths: Final data for 2013. [http://www.cdc.gov/nchs/data/nvsr/nvsr64/nvsr64\\_02.pdf](http://www.cdc.gov/nchs/data/nvsr/nvsr64/nvsr64_02.pdf). October 20, 2015.

h. Centers for Disease Control and Prevention. Injury prevention & control: Data & statistics (WISQARS). <http://www.cdc.gov/injury/wisqars/index.html>. Updated July 13, 2015. October 20, 2015.

**3.81 times more likely** to have a condition identified through newborn screening **than to be injured in a car crash** before age 1



You wouldn't let a child ride home in a car without a car seat...

...so why would you let a child go home without completing newborn screening?



# What to do if a parent would like to refuse?

- According to state law a parent can refuse for any reason
- It should be an informed decision with a clear understanding of the substantial risks
- Provide clear information about newborn screening and its purpose and benefits
- ND requires completion of a refusal documentation form
- The birthing center (if using one) may also require paperwork
- Try to understand why the parent wishes to refuse (common reasons and possible responses on the next slides)

# Responding to Common Parental Concerns with Newborn Screening



# Believe the test will be painful for the baby

- The hearing test and CCHD pulse ox are painless and the baby can sleep through the test
- Some babies sleep through the heel prick as well; can nurse while sample is taken
- Will not leave a scar and heals very quickly



# Tests are too expensive

- Nearly all insurance plans should cover it as does ND / MN Medicaid
- Even if not covered by insurance, the out-of-pocket cost of \$75 is a great price, particularly considering the services provided
- Commercial screening, in comparison, can run many hundreds of dollars

# Worried about a false-positive

- False positive screens or inconclusive screens do occur
- Much more likely to have a false positive than a true positive (have to set the test criteria very cautiously to be sure no babies with the condition are missed)
- How this is handled by the health provider can make a big difference in the parents' response; terminology and clarity of communication critical as is pre-screening education
- Still better to have a false-positive than to have a condition that is missed

# Storage and use of bloodspot cards

- Used to improve the accuracy of the laboratory's equipment
- Used for research to develop future newborn screening tests or related research
- Babies' rights are carefully protected by law and through rigorous research processes
- NO database of DNA, NO sharing of information with insurance companies (such use would be illegal anyway), NO other use of bloodspot cards
- If still have concerns, visit with the ND Newborn Screening Program – may be able to have card destroyed or returned to the family after the screening is completed

# Summary

- Refusal of newborn screening represents a significant risk to the health of the baby
- Risk is much higher with expanded newborn screening than it once was when only PKU was screened
- Clear communication between a trusted medical professional and a parent is the best way to support a decision in favor of newborn screening
- If a parent wants to refuse, be sure they understand the risks and offer to address concerns as possible, be sure to use appropriate documentation forms
  - When in doubt, contact the North Dakota Newborn Screening Program for help!